Cystic Fibrosis (CF)

Primary Defect

A defective protein disrupts the movement of salt and water across cell membranes. A primary effect of this is accumulation of thick mucus that interferes with lung and digestive function.

Screening Test

Screening involves measuring an enzyme produced by the pancreas (immunoreactive trypsinogen or IRT). Diagnosis is established by measuring salt levels in sweat (sweat test) or testing DNA. Newborn screening reduces average age at diagnoses by 14 months. In Washington, screening will fail to detect one affected child (false negative) for every fifty that are found with cystic fibrosis.

Cause & Frequency

Cystic fibrosis is an inherited disorder that occurs when each parent passes on a faulty gene for the protein involved in moving salt and water across cell membranes. One defect (called delta F508) is most common in Northern Europeans and is associated with severe clinical outcomes. Over a thousand other defects have been identified, some associated with more mild disease states. There is significant clinical variability even among those with identical genetic defects. Occurrence is highly variable among different populations. About 1 in 2,000 Northern European, 1 in 17,000 African American and 1 in 9,000 Hispanic infants are affected. In Washington, about 25 children are expected to be born with cystic fibrosis each year.

If Untreated

Cystic fibrosis has a substantial impact on lung function and leads to lung infections and severe malnutrition with associated consequences due to abnormal production of digestive enzymes and poor uptake of essential vitamins. Other features include liver disease, abnormal glucose tolerance, and infertility.

Treatment

Treatments are aimed primarily at maintaining lung function, preventing infections, and enhancing nutritional status through measures such as enzyme and vitamin supplements.

With Treatment

Improved treatments have dramatically increased life expectancy from approximately three years in the 1950s to the late 30s today. Early detection and treatment through newborn screening can result in significantly improved nourishment, improved learning, increased survival and reduced use of health care. The impact of early detection on lung function is less certain: some studies have shown significant improvement, while others have not.